Diagnosis Service Report

[redacted]
Patient

[redacted]
Sex

Social Security Number

[redacted] F 000000000

Whole Blood
Test Ordered

Not Available
Test Requested

Spinal Muscular Atrophy DNA Test

Requesting Physician

Dr. Ostrer

Report to

Rhode Island Hospital

Address

Central Coll. / RM-201 / SW Pavi

593 Eddy Street

Providence, RI 02903

Additional Address to

Institution Number

[redacted]
Number

Patient Number

[redacted]

07/01/2003
Accession Date

07/03/2003
Report Date

07/17/2003

Interpretation

This individual does not possess a deletion of either exons 7 or 8 in the survival motor neuron (SMN) gene and therefore this test fails to confirm the diagnosis of Spinal Muscular Atrophy (SMA).

Technical Results

SMN exon 7: no deletions detected
SMN exon 8: no deletions detected

Methods

Direct testing for deletions of exon 7 and /or 8 in the SMN gene was performed by PCR + amplification and restriction endonuclease digestion of genomic DNA [1,2]. This analysis, as performed here, is greater than 99% accurate.

This test is performed pursuant to a PCR license agreement with Roche Molecular Systems, Inc.
This testing service is covered by a pending patent.

Comments

This analysis did not identify deletions of either exons 7 or 8 in the SMN gene. Detectable deletions of exons 7 and/or exon 8 of SMN have been reported to occur with a frequency ranging from 84% to 100% of patients with clinically diagnosed SMA. [3] Therefore, this result fails to confirm the diagnosis of SMA but does not exclude the diagnosis of this disease due to mutations not detected or recognized at this or other loci.

References

1. Lefebvre, S., et al. (1995) Cell 80: 155-165
2. Van der Steege G., et al. (1995) Lancet 345: 985-986
3. Crawford, T.O., Pardo C.A. (1996) Neurobiol Dis 3: 97-110
4. Cobben, J.M. (1995) Am J Hum Mol Genet 57: 805-808
5. Hahnen E., et al. (1995) Hum Mol Genet 4: 1927-1933
6. Brahe, C., et al. (1995) Am J Med Genet 59: 101-102

*** *FINAL REPORT* *** ver 1.0

This test was developed and its performance characteristics determined by Athena Diagnostics, Inc. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes and should not be regarded as investigational or for research only. Athena Diagnostics is licensed under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) to perform high complexity clinical testing. Athena Diagnostics has performed assay validation studies and has developed its laboratory protocols and operating procedures in consultation with experts in the field and in accordance with the standards of the National Committee on Clinical Laboratory Standards (NCCLS).

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